The London Conference on "The Normal Human Karyotype"*

28th – 30th August, 1963

The present meeting was called to consider developments since the Denver Conference and to assess the degree to which these may have aided characterization of individual chromosomes or may have revealed inadequacies in the earlier identifications. There was general agreement that the seven groups proposed in the Denver numbering system have stood the test of time, and that little or no uncertainty exists in the group assignment of any chromosome. However within certain groups identification of individual chromosomes is questionable. The groups are sometimes designated by letters, such as A to G, as well as by the numbering scheme described in the Denver classification.

A comparison of data submitted by different investigators revelaed that surprisingly good agreement on relative chromosomal length and arm ratios or centromeric indices can be obtained in different laboratories provided that mitotic figures, clearly delineated and free from irregularities of condensation pattern, are selected for analysis. The dispersion of chromosomal measurements appears to be due to both biological and technical factors and, while some of these variations are random, others appear to be systematically produced, as in the effect of exposure to colchicine which may contract some chromosomes more than others. It is hoped that specific procedures maximizing ease of identification will soon be forthcoming.

The new techniques considered included the demonstration of specific chromosomal constrictions and autoradiographic determination of the time at which H^3 thymidine is incorporated in specific chromosomes and chromosomal regions. Both of these methods contribut to the characterization of specific chromosomes. However variability exists in techniques and material; so that at present neither method permits absolute identification of chromosomes. There was discussion on the use of methods for DNA measurement in each chromosome arm, on electron microscopy, on satellite association, on specific location of certain chromosomes within spread mitotic figures, on physicochemical separation of chromosomes in suspension, on the use of reagents which may characterize chromosome segments and on the ash patterns produced by micro-incineration.

It was agreed at Denver that the autosomes should be serially numbered 1 to 22 as nearly as possible in descending order of length. Identification of individual chromosomes is based on size, position of centromere and other morphological features. The sex chromosomes are referred to as X and Y.

The most important newly recognized features are so-called secondary constrictions** and differential patterns of incorporation of isotopically labelled thymidine in specific chromosomes as demonstrated by autoradiography. Secondary constrictions are only seen in certain types of preparations and then in a fraction of the cells.

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** Apart from satellite stalks the term 'secondary constriction' connotes achromatic regions which may appear as attenuated parts.

Variability is observable in the morphology of certain chromosomes. This is especially well established for the Y chromosomes in which great differences in size occur and which are heritable from father to son. Heritable polymorphism has also been observed in satellited chromosomes and probably in No. 16.

Specific Comments

It is emphasized that assignment of numbers to chromosome pairs on the basis of the morphological criteria here discussed includes an element of arbitrariness; it does not ensure homology.

Group 1–3 (A). No. 1. In many cells a secondary constriction is observed in the proximal region of the long arm.

Group 4–5 (B). No additional information is available which can be used to distinguish these two chromosome pairs.

Group 6–12 and the X chromosome (C). Of the autosomes in this group, four are comparatively metacentric. It is proposed that they should be numbered 6, 7, 8 and 11. The X chromosome belongs to this sub-group.

Three chromosomes are sub-metacentric. They should be numbered 9, 10 and 12.

A secondary constriction is found in the proximal part of the long arm of at least one of the pairs in the whole group.

In normal female cells, one chromosome characteristically incorporates isotopically labelled thymidine over most of its length later than the others in the group. This chromosome is believed to be an X.

Group 13–15 (D). Satellites have now been detected in all three chromosome pairs.

Group 16–18 (E). A secondary constriction has been frequently seen in the proximal part of the long arm of No. 16.

Group 19–20 (F). No additional information is available which can be used to distinguish these two chromosome pairs.

Group 21–22 and the Y chromosome (G). Satellites have now been detected on both Nos. 21 and 22.

The most common Y is larger than either 21 or 22, its centric constriction is often indistinct and a secondary constriction is frequently seen in the long arm; the terminal region of the long arm may be poorly defined. Typically, the two long arm chromatids of the Y appear to diverge less than those of other chromosomes.

L. S. Penrose, London

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Symposium über «Diurese und Diuretika in Klinik und Therapie». Sekretär der Akademie: Prof. Walter Montorsi, Via Festa del Perdono 3, Mailand.